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Preface

Prenatal testing

Jacob A. Canick, PhD Geralyn M. Lambert-Messerlian, PhD
Guest Editors

"Is my baby normal?" That is the question on the minds of all expectant parents. As health care providers, we are concerned about the baby, and also the mother and the course of her pregnancy. Now, because of the explosion of knowledge in biology and medicine and because of impressive innovations in diagnostic methods, we can provide answers in ways that were barely anticipated just a few decades ago. Thirty years ago, as many as 12 of the 14 topics in prenatal testing that make up this issue of the *Clinics in Laboratory Medicine* were not yet part of prenatal care.

This issue is devoted to those screening and diagnostic procedures that can identify serious birth defects and various obstetrical complications. As is true in most areas of medicine, such procedures involve both laboratory and clinical techniques. In fact, specific areas of prenatal testing have been at the forefront of integrating laboratory and clinical data into comprehensive reports. Examples of the power of combined assessment of prenatal conditions include the use of ultrasound findings in conjunction with analyte measurement in testing for chromosomal abnormalities, early pregnancy loss, and infectious disease. To accentuate the growing interaction between the laboratory and clinical diagnostic disciplines, each topic in this issue is approached from both perspectives whenever possible. We hope that readers find this approach innovative and helpful.

The first prenatal test is the assessment of early pregnancy viability. Measurement of human chorionic gonadotropin (hCG) has served as the basis for early pregnancy detection since the first rabbit was injected with pregnancy urine. Dr. Cole and colleagues provide an up-to-the-minute

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description of how the biochemistry of hCG has helped in the development of more specific tests for early pregnancy and its disorders. Recurrent pregnancy loss can sometimes be attributed to antiphospholipid syndromes; Drs. Rote and Stetzer provide both clinical and biological data in addition to their perspectives on this complex and current topic.

Infections can pose a threat to pregnancy viability at any time. Dr. Andrews and colleagues discuss a number of the infectious diseases that are harmful to a baby both prenatally and in the newborn period and highlight the use of ultrasound as a complement to laboratory testing. One possible repercussion of infection in pregnancy is fetal anemia. The various causes, manifestations, and identification of fetal anemia and other hematologic disorders are described by Drs. Rubin and Hansen, who give special attention to recent molecular diagnostic methods.

Despite advances in prenatal care, almost 10% of all babies are premature (born before 37 gestational weeks). There are multiple causes of prematurity; Dr. Lockwood categorizes them and describes new tests that can predict preterm delivery. One of the most serious consequences of preterm delivery is fetal respiratory distress syndrome. Dr. Torday provides a historical perspective on the subject of testing for fetal lung maturation and compares the various laboratory methods that have been proposed and implemented.

The first screening test to be offered to all pregnant women was maternal serum AFP measurement, which was used for the detection of neural tube defects. Population-based prenatal screening expanded soon thereafter with the discovery that AFP and, later, other maternal serum analytes were abnormal in Down syndrome pregnancy. We and our colleagues, Drs. Bombard and Saller and Mr. Kellner, describe these screening methods. Serum markers in Down syndrome screening are known to be associated with other adverse pregnancy outcomes. Recent data suggest that a test combining certain of the serum markers used in Down syndrome screening may be useful in predicting pre-eclampsia. In fact, optimal test performance in screening for Down syndrome and pre-eclampsia will almost certainly involve the use of serum and ultrasound markers together. Drs. Lyall and Giudice have collaborated with us to describe both diagnostic and predictive tests for pre-eclampsia.

Diabetes in pregnancy remains a management challenge to the obstetrician. Historical and clinical risk factors are not adequate to identify patients who will be affected. Drs. Coustan and Carpenter from Women and Infants' Hospital were among the first to recommend that prenatal screening for gestational diabetes be offered to all women. In the article by Drs. Carpenter and O'Brien, the most recent advances in screening methods to identify gestational diabetics are described.

No current review of laboratory diagnostic methods would be complete without descriptions of molecular and cytogenetic testing. Drs. Donnenfeld and Lamb present a timely discussion on cytogenetic and molecular cytogenetic methods, as well as the latest in prenatal specimen collection. Drs.

Tantravahi and Wheeler review DNA-based methods used in the prenatal diagnosis of a range of inherited diseases, describing both Mendelian and non-Mendelian disorders. In contrast, Drs. Richards and Haddow focus on prenatal screening for cystic fibrosis, an inherited disease for which testing has been recently recommended for all pregnant women by the American College of Obstetricians and Gynecologists and the American College of Medical Genetics.

This issue concludes with a discussion of an innovative method in quality assurance, so-called "epidemiologic monitoring." Dr. Knight and Mr. Palomaki use prenatal screening tests for Down syndrome and neural tube defects as a model to illustrate the method; however, such population-based monitoring should be useful in all laboratory-based testing.

We hope that the information provided in this issue emphasizes the need for close interaction between the laboratory and the practitioner in the management of pregnancy. Although it is impossible to ensure that a baby and a pregnancy will be normal, collaboration between medical disciplines to provide better prenatal tests will ultimately lead to better outcomes.

Jacob A. Canick, PhD
Geralyn M. Lambert-Messerlian, PhD
Division of Prenatal and Special Testing
Department of Pathology and Laboratory Medicine
Brown Medical School
Women and Infants' Hospital
70 Elm Street, 2nd Floor
Providence, RI 02903, USA

E-mail address: gmesserl@wihri.org (G.M. Lambert-Messerlian)